Haplogroup Prediction in the Ghanaian Population using Haplotype data of 27 Yfiler® Plus loci and TaqMan SNP Genotyping

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Abstract

This study describes the use of the 27 loci Yfiler® Plus kit and TaqMan™ SNP genotyping to characterise and predict the haplogroups of Y chromosomes within the four major ethnic populations of Ghana. Haplogroups were assigned using the desktop NevGen software (https://www.nevgen.org/). The E1b1a and E1b1b haplogroups are the most common in the Ghanaian population and form 95% of the dataset. The Mole-Dagomba sub-population had 4. 8% assigned to the haplogroups G, H, R1b, R2 and T. The Ewe had two samples assigned to haplogroups C and D whilst the Akan had one sample each assigned to haplogroups B, J1 and J2. The NevGen predicted haplogroups were further screened with TaqMan™ genotyping for confirmation. In conclusion, ≈95% of the dataset was classified as M-E1b1a using NevGen combined with TaqMan™ SNP Genotyping for confirmation. The TaqMan™ also revealed 5% as J1 and other haplogroups, using an in-house control from the J1 haplogroup.

1. Introduction

Few forensic genetic studies have been conducted in sub-Saharan Africa. Presently, limited forensic genetic studies have been conducted in Ghana to characterise the population structure [1]. This study describes the use of the 27 loci Yfiler® Plus kit (Thermo Fisher Scientific, 2016) and TaqMan™ SNP genotyping to characterise and predict the haplogroup distribution in the four major ethnic populations of Ghana (Akan, Ewe, Ga-Adangbe and Mole-Dagomba). The dataset passed the quality checks of the Y-chromosome STR Haplotype Reference Database (YHRD) and is available from YHRD, release 62 (YA004641-Akan; YA004643-Ewe; YA004644-Ga-Adangbe and YA004645-Mole-Dagomba).

2. Materials and Methods

2.1 DNA Collection, purification, and quantification

Samples from 588 male unrelated Ghanaians were collected with informed consent from the four major ethnic groups in Ghana (Akan, Mole-Dagomba, Ewe and Ga-Dangbe), with about 98.4% ethno-geographic coverage. Genomic DNA was extracted with Gentra Puregene (Qiagen® UK) and quantified with the high sensitivity dsDNA Qubit kit (Thermo Fisher Scientific, USA) using the Qubit 3.0 Fluorometer following the manufacturers protocol.

2.2 Amplification and fragment detection

Amplification was performed using the Yfiler™ Plus Kit (Thermo Fisher Scientific, 2016) according to the manufacturer's standard protocol but a modified quarter reaction volume to generate Y-haplotype data for 27 Y-STR loci.

2.3 TaqMan™ SNP genotyping

The SNP Genotyping was carried out using the TaqPath™ ProAmp Master Mix and the 20X TaqMan™ SNP Genotyping probes (Thermo Fisher Scientific) using the QuanStudio™ 5 real-time thermal cycler. The reaction was performed following the manufacturer's guidelines, with a 12.25 µl reaction volume. The allelic discrimination was carried out using the custom QuantStudio™ Design and Analysis Desktop Software v1.4.3. The post-read temperature of 60 °C was used for the analysis.

2.4. Data analysis

The allelic discrimination plots and the raw data in Excel were exported for further analysis. The Y haplogroups were predicted from the Y-STR data with the online haplogroup predictor NevGen (http://www.nevgen.org/), which uses the Bayesian approach [2]. All data were presented as mean ± standard deviation. A p -value= < 0.05 was taken as significant.

3. Results and discussion

The Y-STR haplogroup prediction and TaqMan™ SNP genotyping screening (Table 1) reported >95% E1b1a and E1b1b haplogroups assignment to the four Ghanaian subpopulations.

Table 1 Haplogroup prediction for 588 samples of the four Ghanaian subpopulations in this study. UP: unpredicted haplogroup.

Haplogroup Predictions																		
pop	n	A	B	C	D	E1b1a	E1b1b	G	H	I2	J1	J2	Q	R1a	R1b	R2	T	UP
All	588	6	1	1	1	553	9	1	2	2	1	3			2	1	2	3
Akan	182	3	1			171	3			1	1							2
Ewe	209	1		1	1	201	4					1						
Ga-Adangbe	32	1				30	0					1						
Mole-Dagomba	165	1				151	2	1	2	1		1			2	1	2	1

The slightly higher level of heterogeneity in the Mole-Dagomba could be due to different population groups who have left a genetic mark in the geographical area following the trans-Sahara trade in the 13th century; the Mole-Dagomba are understood to be related to the Mossi kingdoms of Burkina Faso [3] and the presence of Arabic influence is evident in their religion (Islam) and culture. The presence of haplogroup J, which is associated with the Middle East and parts of Europe, in the Mole-Dagomba, provides further evidence of the influence of the Middle Eastern population on the Mole-Dagomba [4].

Similar anthropological studies in Ghana have reported the E1b1a and E1b1b as the main haplogroups of the Bimoba tribe in north-eastern Ghana [5]. Other African studies have reported genetic differentiation between ethnic groups separated by political borders. One of such studies, Fortes-Lima, *et al.,* [6] reported a genetic differentiation between the Nigerian and Beninese Yoruba. The researchers genotyped 288 male samples from Benin and Ivory Coast using the Yfiler™ kit and compared the results to published data from the region. They also reported the E1b1a1-M2 as the predominant haplogroup.

Many population studies of the Y-chromosome from the major language families; Niger-Congo, Nilo-Saharan, Khoisan and Afroasiatic in the West African subregion reported E1b1a and E1b1b haplogroups as the dominant groups although other haplogroups, occurring at a minor frequency were reported [7]. The inferred haplogroup E1b1a is common with the Bantu language speakers, who migrated from Benue River Valley, between Nigeria and Cameroon [8,9]. Studies of the Y-chromosome involving the geographic neighbours of Ghana with same

ethnic groups across the borders have reported haplogroups consistent with this study [10,11].

4. Conclusions

The results of this study present a study of Y chromosome haplotypes in Ghana, samples were selected randomly from broad ethnic groups and profiled using commonly used autosomal and Y chromosome forensic markers [1]. The limited haplogroup diversity renders the haplogrouping of casework samples of limited value as it will add very little to the discriminatory levels obtained with STR markers.

Declaration of Competing Interest

None

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